# MISSOURI NEWBORN SCREENING

## 2016 Annual Report


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Missouri Department of Health and Senior Services

# Acknowledgments

The Missouri State Genetic Advisory Committee and its ancillary Newborn Screening Standing Committee, Sickle Cell Standing Committee, Cystic Fibrosis Standing Committee, Newborn Hearing Screening Standing Committee, and the Lysosomal Storage Disorder Task Force Committee play a vital role in supporting the activities of the Missouri Department of Health and Senior Services Newborn Screening Program.

The expertise the committees provide is complemented by department staff who are dedicated to helping Missouri children receive the best care available when diagnosed with one of the serious medical conditions detectable through screening tests.

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Missouri Department of Health and Senior Services
Division of Community and Public Health
Section for Healthy Families and Youth
Bureau of Genetics and Healthy Childhood
and
Missouri State Public Health Laboratory

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# Screening Spotlight: Newborn Screening Laboratory Converts to a Six Day Work Week

In 2016, the Missouri State Public Health Laboratory (MSPHL) achieved a paramount improvement in its newborn screening system by adding Saturdays and most holidays to the newborn screening testing regimen. The only holidays during which the Newborn Screening Laboratory (NBSL) does not test specimens are Thanksgiving, Christmas Day, and New Year's Day. Prior to this change, the NBSL tested newborn screening specimens Monday through Friday and did not test on any state holidays.

This change in testing regimen was not an easy task for a laboratory unit that tests for over six million analytes per year with ten different testing methodologies. The addition of testing on Saturdays and most holidays came with several challenges, including: funding to hire staff, hiring staff willing to work on Saturdays and holidays, and making the necessary enhancements to the current courier system. Due to legislative funding headed by a State Representative from the Kansas City area, the MSPHL was able to develop a systematic plan to overcome these challenges and successfully implement the necessary changes.

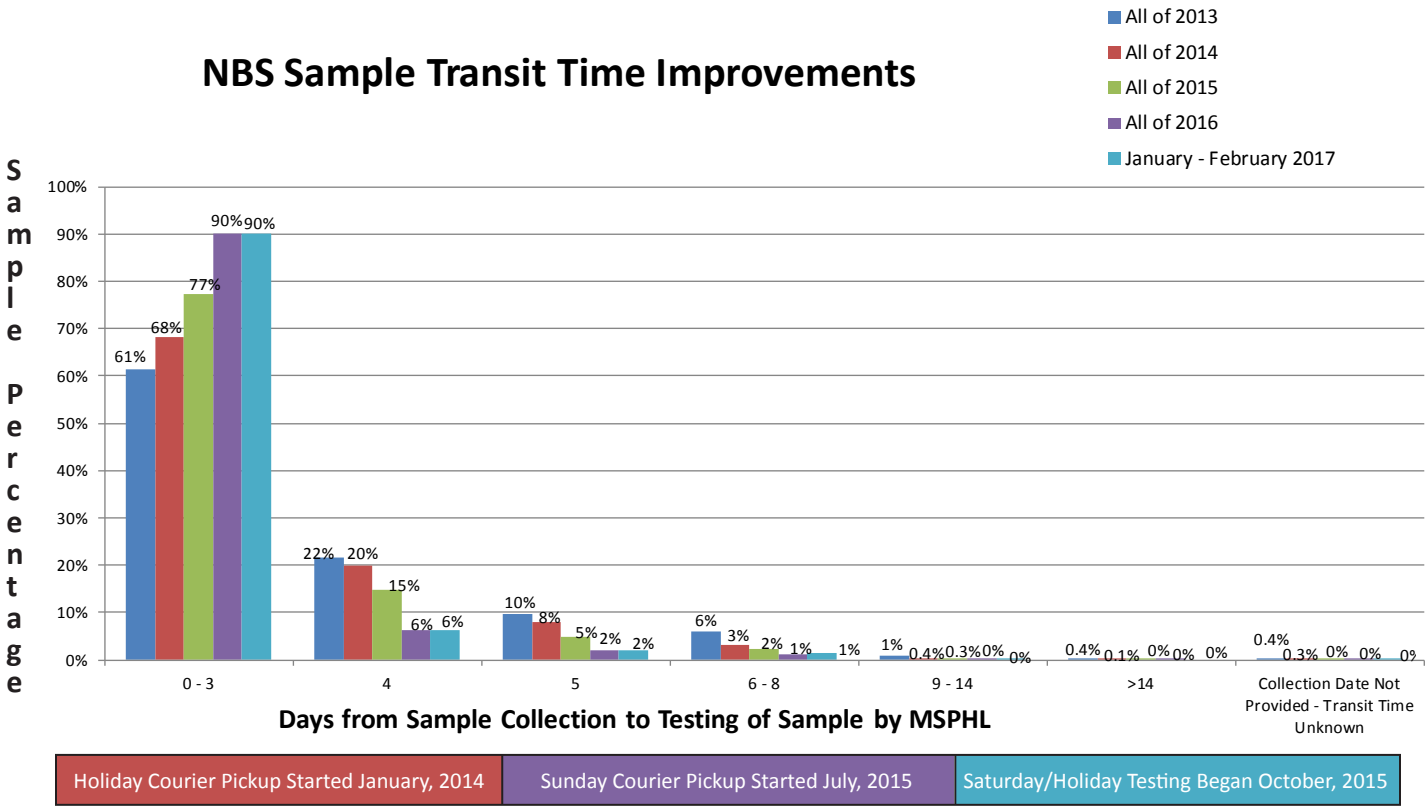
The first step was to enhance the courier system by adding holiday and Sunday pickups at the birthing hospitals. In order to maximize efficiency, the courier needs to pick up samples one day ahead of the NBSL testing, so beginning July 5, 2015 couriers began picking up newborn screening samples Sundays through Fridays, including holidays (see Missouri NBS Sample Transit Time Improvements graph). In addition, eight birthing hospitals that were not formerly receiving courier services were added to the pick up routes. These enhancements provided a 17 percent increase in the number of samples received by the NBSL within 3 days of collection at the hospital.

*This staff of approximately 18 employees, led by a fulltime laboratory manager, became known as the "Weekend Warriors."*

After the courier enhancements were in place, Saturday and holiday testing could begin. The goal for staffing was to maintain a voluntary process where the current NBSL staff would not be forced to rotate Saturday and holiday coverage. Utilizing an employment status called "secondary assignment" made this possible. The NBSL was able to hire current MSPHL scientists through secondary assignment, also known as Hourly & Intermittent (H&I's), who wanted to work some Saturdays and holidays for additional pay. This staff of approximately 18 employees, led by a fulltime laboratory manager, became known as the "Weekend Warriors." The Saturday and holiday testing began on October 3, 2015 and the MSPHL immediately realized an additional 9 percent increase in samples tested within 3 days of collection, bringing the total timeliness improvement to about 28 percent.

As a result of the NBSL work expansion the MSPHL has realized several valuable benefits, including:

- Newborn screening timeliness has improved providing a much better turnaround time for the detection of many time-critical disorders.
- A collaborative workforce has been established between the regular NBSL staff and the Weekend Warrior team.
- The work expansion has assisted in breaking down silos between laboratory units as the Weekend Warriors come from several units throughout the laboratory.
- The Weekend Warrior team has developed an environment of comradery.
- The Saturday and holiday testing has reduced the huge surge days (some as high as 900 samples) that resulted from weekends and three-day weekends, resulting in a more even workflow for all NBSL staff.



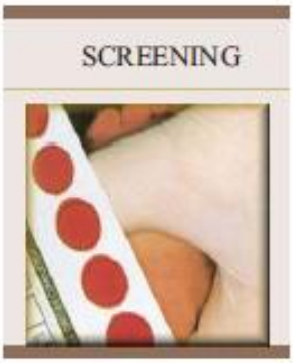

Note: Holiday courier pickup was added in January of 2014. The Sunday courier addition, along with eight additional hospital pickup sites, was implemented July 5, 2015. Saturday and holiday testing began October 3, 2015.

When the sample collection date is not provided, the sample is tested for everything, however, we cannot provide normal results for all testing categories. A repeat screen is required.

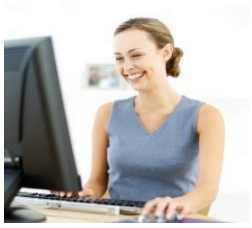
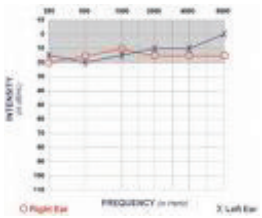
As improving timeliness continues to be a focus in newborn screening, state newborn screening laboratories must continuously evaluate their processes. This has resulted in an increase of state newborn screening laboratories expanding their work week throughout the nation. Currently, about half of state newborn screening laboratories test fully on Saturdays. Missouri's volunteer model has been inquired about by several other states interested in implementing a similar process. This effort has been a tremendous success, and the Newborn Screening Program would like to thank all those who have and are still contributing to this remarkable improvement.

# The Weekend Warriors

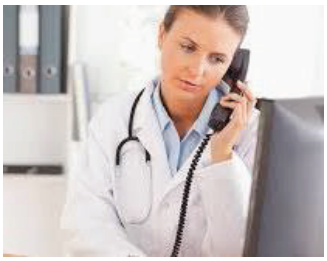
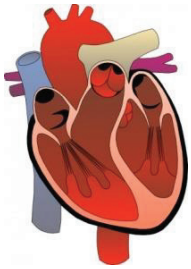
# The Newborn Screening Process

| 1: TESTING | 2: FOLLOW-UP | 3: DIAGNOSIS/ INTERVENTION | 4: TREATMENT & MANAGEMENT |
|---|---|---|---|
| The baby's heel is pricked and a few drops of blood are collected on a filter paper 24 to 48 hours after birth. | Positive screen results are reported by phone/ fax/letter from lab and follow-up staff to baby's physician. Results are also sent to the appropriate Genetic Tertiary Center in Missouri for follow-up. | Depending on the screen result and the condition screened, repeat or confirmatory testing occurs at the genetic center. | Once diagnosis is made, treatment begins. For some life-threatening conditions, treatment may occur prior to diagnosis - on the recommendation of a specialist. |
|  |  |  |  |
| The dried blood spot specimen is shipped to the State Public Health Laboratory. | Baby's physician or health care provider contacts baby's parents. | Parent education for signs/symptoms to watch for is conducted. | Parents receive treatment guidelines/ education. Team support services, as appropriate, include:<br>- Metabolic dietitian monitoring and consultation<br>- Ongoing blood monitoring<br>- Referral to early intervention services<br>- Pulmonary/CF services<br>- Pediatric endocrine monitoring<br>- Pediatric hematology monitoring<br>- Genetic counseling and consideration of family testing<br>- Other allied health services as needed |
|  |  |  | |
| Specimen is tested for multiple conditions. | Parents bring baby back in for evaluation and more testing at the genetic center. | Baby's physician consults with the specialist appropriate to the condition. | |
| | |  | |

# The Newborn Hearing Screening Process

| 1: SCREENING | 2: FOLLOW-UP | 3: EVALUATION | 4: INTERVENTION |
|---|---|---|---|
| Baby is born.<br>Hospital screens for hearing loss and checks for risk factors for late onset hearing loss prior to discharge.<br>  <br>Hospital submits results to the Missouri Department of Health and Senior Services (DHSS) via the Missouri Electronic Vital Records (MoEVR) system or on a paper form.<br>  <br>Manual data is entered into the Missouri Health Strategic Architectures and Information Cooperative (MOHSAIC) data system.<br>DHSS retrieves results from the MOHSAIC data system.<br>  | Hospital reports results to parents and baby's physician.<br>  <br>DHSS sends letters to parents and physicians of newborns who did not pass or who missed the screening.<br>  <br>Parents return baby to hospital/health care provider 1-3 weeks after initial referral.<br>  | Audiologist evaluates babies that don't pass a hearing screening by 3 months of age.<br>  <br>Audiologist reports evaluation results to DHSS.<br>  <br>Audiologist identifies risk factors and makes recommendations.<br>  <br>DHSS sends letters to the families of children diagnosed with permanent hearing loss and refers to Missouri's Part C of the Individuals with Disabilities Education Act (IDEA) program, First Steps. | Babies diagnosed with permanent hearing loss enroll in First Steps (early intervention service) by 6 months of age.<br>Missouri First steps EARLY INTERVENTION<br>Babies receive services from the following as appropriate: Primary Care Physician, Otolaryngologist, Geneticist, and Ophthalmologist.<br>  <br>Baby may be a candidate for: Hearing aids, cochlear implant, sign language instruction, or speech and language services.<br>  |

# The Newborn Critical Congenital Heart Disease Screening Process

| 1: SCREENING | 2: FOLLOW-UP | 3: EVALUATION | 4: INTERVENTION |
|---|---|---|---|
| Baby is born.<br>  <br>Hospital or midwife screens for critical congenital heart disease (CCHD) between 24 and 48 hours after birth or prior to discharge.<br><br>Screening should be in accordance with the American Academy of Pediatrics and American Heart Association guidelines.<br>  <br>Screening should be done while baby is warm, calm, and awake.<br>  | If screening is normal, no further action is necessary.<br><br>If baby does not pass the screening, further evaluation will be necessary and the primary care provider should be contacted as soon as possible.<br>  | The baby's primary care provider will perform a thorough physical examination to rule out any non-cardiac issues that may have prevented baby from passing the CCHD screen.<br>  <br>An echocardiogram may be done to look for a CCHD.<br>  <br>The echocardiogram should be read by a pediatric cardiologist.<br>  | Babies diagnosed with CCHDs will typically require surgical or catheter intervention within the first year of life.<br>  <br>Parents will receive treatment guidelines and education.<br>  <br>Babies may receive services from the following as appropriate: primary care provider, pediatric cardiologist, geneticist, nurse, nutrionist, pharmacist, social worker, and child life specialist.<br>  |

# Newborn Screening Contact Information

## Telephone Contacts:

| | |
|---|---|
| Newborn Screening Laboratory main number | 573-751-2662 |
| Order newborn screening specimen forms | 573-751-3334 |
| Genetics and Healthy Childhood, for follow-up information | 800-877-6246 |

## Web Addresses:

Critical Congenital Heart Disease – www.health.mo.gov/cchd

Newborn Screening Laboratory – http://health.mo.gov/lab/newborn/

Newborn Screening Program – www.health.mo.gov/newbornscreening

Newborn Hearing Screening Program – www.health.mo.gov/newbornscreening

## Appendix 1: Disorders Confirmed for 2016 and Projected Incidence Rates

| DISORDER | DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE | PROJECTED INCIDENCE RATE |
|---|---|---|
| **Amino Acid Disorders** | ***12*** | **1/6,500*** |
| Argininemia | | |
| Argininosuccinic acidemia | 2 | |
| Citrullinemia type I | 1 | |
| Citrullinemia type II | | |
| Defects of biopterin cofactor biosynthesis | | |
| Defects of biopterin cofactor regeneration | | |
| Homocystinuria | | |
| Hypermethioninemia | | |
| Hyperphenylalaninemia | | |
| Hyperphenylalaninemia, benign | | |
| Maple syrup urine disease | | |
| Maternal PKU | | |
| Phenylketonuria (PKU) | 9 | |
| Tyrosinemia type I | | |
| Tyrosinemia type II | | |
| Tyrosinemia type III | | |
| **Biotinidase Deficiency (BIOT)** | ***13*** | **1/6,000*** |
| Partial biotinidase deficiency | 10 | |
| Profound biotinidase deficiency | 3 | |
| **Congenital Adrenal Hyperplasia (CAH)** | ***4*** | **1/19,500** |
| Congenital adrenal hyperplasia non salt waster | | |
| Congenital adrenal hyperplasia salt waster | 4 | |
| **Congenital Primary Hypothyroidism (CH)** | ***45*** | **1/1,800** |
| **Cystic Fibrosis (CF)** | ***26*** | **1/3,000** |
| Cystic fibrosis | 21 | |
| Cystic fibrosis transmembrane conductance regulator (CRTR) - related metabolic syndrome (CRMS) | 5 | |
| **Fatty Acid Oxidation Disorders** | ***25*** | **1/3,400*** |
| Carnitine acylcarnitine translocase deficiency | | |
| Carnitine uptake deficiency | | |
| Carnitine palmitoyl transferase deficiency I | | |
| Carnitine palmitoyl transferase deficiency II | | |
| Dienoyl-CoA reductase deficiency | | |
| Glutaric acidemia type II | | |

| DISORDER | DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE | PROJECTED INCIDENCE RATE |
|---|---|---|
| Isobutyryl-CoA dehydrogenase deficiency | 1 | |
| Long-chain hydroxyacyl-CoA dehydrogenase deficiency | 1 | |
| Maternal carnitine uptake deficiency | | |
| Medium-chain acyl-CoA dehydrogenase deficiency | 9 | |
| Medium-chain ketoacyl-CoA thiolase deficiency | | |
| Medium/Short chain L-3 hydroxy acyl-CoA dehydrogenase deficiency | | |
| Short-chain acyl-CoA dehydrogenase deficiency | 5 | |
| Trifunctional protein deficiency | | |
| Very-long chain acyl-CoA dehydrogenase deficiency | 9 | |
| **Galactosemia (GALT)** | ***11*** | **1/7,000*** |
| Classical galactosemia | 3 | **1/26,000**** |
| Duarte galactosemia | 8 | |
| **Lysosomal Storage Disorders (LSD)** | ***37*** | **1/2,100*** |
| **Fabry Disease** | **21** | |
| Fabry | 20 | |
| Unknown onset | | |
| Genotype of unknown significance | 1 | |
| **Gaucher Disease** | **2** | |
| Gaucher type 1 (non-neuropathic) | 1 | |
| Unknown onset | | |
| Genotype of unknown significance | 1 | |
| **Hurler Syndrome** | **2** | |
| Attentuated | | |
| Severe | | |
| Genotype of unknown significance | 2 | |
| **Krabbe Disease** | **2** | |
| Infantile onset | 1 | |
| Later onset | | |
| Unknown onset | 1 | |
| Genotype of unknown significance | | |
| **Pompe Disease** | **10** | |
| Classical infantile o nset | 2 | |
| Non-classical infantile onset | | |
| Later onset | 7 | |

| DISORDER | DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE | PROJECTED INCIDENCE RATE |
|---|---|---|
| Unknown onset | | |
| Genotype of unknown significance | 1 | |
| **Organic Acid Disorders** | *7* | **1/11,100*** |
| 2-Methyl-3-hydroxybutyric aciduria | | |
| 2-Methylbutyryl-CoA dehydrogenase deficiency | 1 | |
| 3-Hydroxy-3-methylglutaric aciduria | | |
| 3-Methylcrotonyl-CoA carboxylase deficiency | | |
| 3-Methylglutaconic aciduria | | |
| Beta ketothiolase | | |
| Glutaric acidemia, type I | 1 | |
| Isobutyryl-CoA dehydrogenase deficiency | | |
| Isovaleric acidemia | | |
| Malonic acidemia | | |
| Methylmalonic acidemia (CBL A,B; vitamin B12 disorders) | | |
| Methylmalonic acidemia (CBL, C,D) | 1 | |
| Methylmalonic acidemia (MUT, methylmalonyl-CoA mutase) | | |
| Multiple carboxylase deficiency | | |
| Propionic acidemia | 2 | |
| Forminioglutamic acid (FIGLU) not a disorder on the newborn screening panel but is found | 2 | |
| **Hemoglobinopathies** | ***35*** | **1/2,200** |
| Sickle cell anemia disease (FS) | 17 | **1/4,500** Total population<br>**1/400** African-American population |
| Sickle hemoglobin-C disease (FSC) | 6 | |
| Sickle beta zero thalassemia disease | | |
| Sickle beta plus thalassemia disease (FSA) | 5 | |
| Sickle hemoglobin-D disease | | |
| Sickle hemoglobin-E disease | | |
| Sickle hemoglobin-O-Arab disease | | |
| Sickle hemoglobin Lepore Boston disease | | |
| Sickle hereditary persistence of fetal hemoglobin (HPFH) disorder | | |
| Sickle “Unidentified” | | |
| Homozygous-C disease (FC) | 2 | |
| Hemoglobin-C beta zero thalassemia disease | | |

| DISORDER | DIAGNOSIS CONFIRMED AS POSITIVE AND UNDER MEDICAL CARE | PROJECTED INCIDENCE RATE |
|---|---|---|
| Hemoglobin-C beta plus thalassemia disease (FCA) | 1 | |
| Hemoglobin-C with unidentified hemoglobin (FCX) | 1 | |
| Homozygous-E disorder | | |
| Hemoglobin-E beta zero thalassemia disease | | |
| Hemoglobin-E beta plus thalassemia disease (FE) | 2 | |
| Homozygous beta zero thalassemia disease | | |
| Double heterozygous beta thalassemia disease | | |
| Hemoglobin-H disease (Highly Elevated Barts) | 1 | |
| Other (FSX) compound heterozygous Hb S and G-Taipei | | |
| Other Disease Condition | | |

*Combined incidence of all disorders in this category
**Incidence only for classical galactosemia

# Appendix 2: Newborn Screening Laboratory Report - Samples Received 2016

| | Newborn Specimens Received | | | |
|---|---|---|---|---|
| | **Initial** | **Repeat** | **Poor Quality** | **Total Infant Specimens** |
| **Jan** | 5,962 | 1,336 | 203 | 7,501 |
| **Feb** | 5,860 | 1,268 | 219 | 7,347 |
| **Mar** | 6,332 | 1,401 | 166 | 7,899 |
| **Apr** | 5,834 | 1,247 | 145 | 7,226 |
| **May** | 6,019 | 1,248 | 136 | 7,403 |
| **Jun** | 6,221 | 1,276 | 157 | 7,654 |
| **Jul** | 6,625 | 1,414 | 183 | 8,222 |
| **Aug** | 6,877 | 1,379 | 179 | 8,435 |
| **Sep** | 6,576 | 1,278 | 140 | 7,994 |
| **Oct** | 6,232 | 1,234 | 158 | 7,624 |
| **Nov** | 5,787 | 1,199 | 200 | 7,186 |
| **Dec** | 6,217 | 1,344 | 242 | 7,803 |
| **Y.T.D.** | **74,542 (80.77%)** | **15,624 (16.93%)** | **2,128 (2.31%)** | **92,294** |

## CALENDAR YEAR 2016

## NEWBORN SCREENING LABORATORY REPORT

## ABNORMAL RESULTS

| Disorder | | Jan | Feb | Mar | Apr | May | Jun | Jul | Aug | Sep | Oct | Nov | Dec | Y.T.D. |
|---|---|---|---|---|---|---|---|---|---|---|---|---|---|---|
| BIO | Confirmed Profound | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 0 | 1 | 1 | **3** |
| | Confirmed Partial | 1 | 1 | 1 | 0 | 2 | 3 | 2 | 0 | 0 | 0 | 0 | 0 | **10** |
| | High Risk | 2 | 2 | 2 | 1 | 2 | 3 | 2 | 1 | 0 | 2 | 1 | 1 | **19** |
| | Borderline Risk | 5 | 2 | 8 | 8 | 8 | 12 | 14 | 7 | 7 | 2 | 3 | 4 | **80** |
| CAH | Confirmed | 0 | 0 | 1 | 1 | 0 | 1 | 1 | 0 | 0 | 0 | 0 | 0 | **4** |
| | High Risk | 2 | 2 | 5 | 3 | 1 | 6 | 3 | 2 | 2 | 6 | 3 | 1 | **36** |
| | Borderline Risk | 53 | 41 | 57 | 59 | 55 | 59 | 50 | 32 | 54 | 47 | 25 | 31 | **563** |
| CF | Confirmed CF | 3 | 3 | 2 | 1 | 1 | 2 | 0 | 1 | 1 | 0 | 4 | 3 | **21** |
| | Confirmed CRMS | 1 | 0 | 1 | 0 | 0 | 0 | 0 | 0 | 0 | 1 | 0 | 1 | **4** |
| | Carriers Identified by NBS | 11 | 14 | 16 | 5 | 15 | 9 | 10 | 4 | 12 | 10 | 5 | 8 | **119** |
| | Referred | 15 | 17 | 20 | 5 | 17 | 11 | 10 | 5 | 13 | 12 | 9 | 11 | **145** |
| | Initial IRT | 74 | 82 | 61 | 47 | 42 | 41 | 52 | 46 | 44 | 56 | 51 | 49 | **645** |
| CH | Confirmed | 6 | 3 | 3 | 4 | 3 | 2 | 4 | 2 | 9 | 4 | 2 | 3 | **45** |
| | High Risk | 8 | 3 | 4 | 5 | 3 | 5 | 4 | 2 | 8 | 6 | 0 | 2 | **50** |
| | Borderline Risk | 102 | 113 | 74 | 65 | 42 | 47 | 44 | 48 | 49 | 75 | 96 | 131 | **886** |
| GALT | Confirmed Classical | 0 | 0 | 0 | 0 | 0 | 0 | 2 | 0 | 0 | 0 | 1 | 0 | **3** |
| | Confirmed DG | 1 | 0 | 3 | 0 | 1 | 0 | 2 | 0 | 1 | 0 | 0 | 0 | **8** |
| | Confirmed G-6-PD | 0 | 0 | 1 | 1 | 0 | 1 | 1 | 0 | 0 | 1 | 0 | 0 | **5** |
| | High Risk | 3 | 0 | 4 | 2 | 1 | 2 | 12 | 0 | 2 | 1 | 1 | 1 | **29** |
| | Borderline Risk | 2 | 2 | 2 | 0 | 0 | 7 | 10 | 5 | 1 | 8 | 3 | 0 | **40** |
| AA | Confirmed | 1 | 1 | 2 | 0 | 0 | 3 | 0 | 0 | 1 | 1 | 1 | 3 | **13** |
| | Referred | 2 | 1 | 2 | 4 | 1 | 2 | 0 | 0 | 2 | 4 | 2 | 3 | **23** |
| | Low Risk | 77 | 70 | 74 | 50 | 58 | 59 | 54 | 56 | 50 | 42 | 33 | 71 | **694** |
| OA | Confirmed | 1 | 0 | 1 | 0 | 2 | 0 | 0 | 1 | 0 | 0 | 2 | 0 | **7** |
| | Referred | 2 | 2 | 2 | 1 | 2 | 0 | 3 | 1 | 2 | 4 | 3 | 2 | **24** |
| | Low Risk | 53 | 49 | 55 | 43 | 49 | 29 | 40 | 42 | 37 | 33 | 46 | 72 | **548** |
| FA | Confirmed | 1 | 2 | 2 | 3 | 3 | 4 | 1 | 0 | 2 | 2 | 1 | 4 | **25** |
| | Referred | 1 | 7 | 6 | 6 | 5 | 9 | 2 | 5 | 2 | 3 | 1 | 6 | **53** |
| | Low Risk | 44 | 41 | 65 | 64 | 52 | 49 | 50 | 56 | 35 | 53 | 59 | 57 | **625** |
| Hb | Sickle Cell Disease | 2 | 4 | 2 | 2 | 4 | 2 | 1 | 2 | 3 | 3 | 1 | 2 | **28** |
| | Other Hemoglobinopathies | 0 | 0 | 0 | 1 | 0 | 0 | 0 | 0 | 2 | 4 | 0 | 0 | **7** |
| | Traits Identified by NBS | 120 | 131 | 134 | 119 | 136 | 131 | 150 | 139 | 153 | 141 | 131 | 158 | **1643** |
| LSD | Confirmed Disorder | 2 | 3 | 2 | 2 | 3 | 5 | 6 | 4 | 3 | 2 | 2 | 3 | **37** |
| | Confirmed Carrier | 0 | 4 | 3 | 3 | 1 | 6 | 2 | 3 | 5 | 1 | 2 | 3 | **33** |
| | Confirmed Pseudo Def. | 3 | 1 | 2 | 2 | 0 | 5 | 4 | 2 | 2 | 1 | 0 | 3 | **25** |
| | High Risk | 5 | 14 | 12 | 13 | 5 | 18 | 18 | 14 | 16 | 4 | 10 | 17 | **146** |
| | Borderline Risk | 14 | 33 | 45 | 48 | 72 | 49 | 70 | 49 | 36 | 55 | 36 | 55 | **562** |
| SCID | Confirmed | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | **0** |
| | Referred | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | **0** |
| | Low Risk | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | 0 | **0** |

**Total Confirmed** **220**

BIO = biotinidase deficiency
CF = cystic fibrosis
GAL = galactosemia
OA = organic acid
Hb = Hemoglobinopathies
CAH = congenital adrenal hyperplasia
CH = congenital hypothyroidism
AA = amino acid
FA = fatty acid
LSD = lysosomal storage disorder
SCID = Severe Combined Immunodeficiency

## Appendix 4: Outcome Data – Newborn Screening Samples and Results

- In 2016, there were 92,294 blood spot samples received in the laboratory. Samples received included:

| Initial | Repeat | Poor Quality |
|---|---|---|
| 74,542 | 15,624 | 2,128 |

- In the process of screening newborns for 70 genetic and metabolic conditions, it is the newborn screening laboratory's role to assess the risk of any abnormal screening by evaluating the marker analytes and the levels that were detected. This risk assessment then dictates different levels of action and follow-up protocols. The 92,294 newborn screening samples received at the state newborn screening laboratory can be separated into two risk categories. The number/percentage of test results falling into these categories during 2016 were:

| High Risk / Referred | Low Risk / Borderline Risk |
|---|---|
| 560 (0.75%) | 4,643 (6.2%) |

**High Risk / Referred** – Results are immediately phoned and faxed to the physician of record and to the contracted genetic referral centers for consultation and confirmatory testing. Final laboratory reports are mailed to the facility that submitted the specimen and the physician of record.

**Low Risk / Borderline Risk** – Final laboratory results are mailed to the physician of record and submitting facility with a comment that a repeat newborn screen is necessary.

- **Two hundred and fifteen (215)** confirmed disorders were diagnosed from these abnormal newborn screen results during 2016.

# Appendix 5: 2016 Poor Quality Samples Report

| | |
|---|---|
| **QUANTITY NOT SUFFICIENT:**<br>Quantity of blood on filter paper not sufficient for testing. Possible causes: removing filter paper before blood has completely filled circle; not allowing an ample size blood drop to form before applying to filter paper; inadequate heel stick procedure. | 409 |
| **INCOMPLETE SATURATION:**<br>Uneven saturation; blood did not soak through the filter paper. Possible causes: removing filter paper before blood has completely filled circle or before blood has soaked through to opposite side; improper capillary tube application; allowing filter paper to come in contact with gloved or ungloved hands or substances such as hand lotion or powder, either before or after blood sample collection. | 519 |
| **SAMPLE ABRADED:**<br>Filter paper scratched, torn or abraded. Possible causes: improper use of capillary tubes. To avoid damaging the filter paper fibers, do not allow the capillary tube to touch the filter paper. Actions such as "coloring in" the circle, repeated dabbing around the circle, or any technique that may scratch, compress, or indent the paper should not be used. | 70 |
| **LAYERED CLOTTED OR SUPERSATURATED:**<br>Possible causes: touching the same circle on filter paper to blood drop several times; filling circle on both sides of filter paper; application of excess blood; clotted swirl marks from improper capillary application. | 873 |
| **DILUTED, DISCOLORED OR CONTAMINATED:**<br>Possible causes: squeezing or milking of area surrounding the puncture site; allowing filter paper to come into contact with gloved or ungloved hands, or substances such as alcohol, formula, antiseptic solutions, water, hand lotion, powder, etc., either before or after blood sample collection; exposing blood spots to direct heat; allowing blood spots to come into contact with tabletop, etc. while drying the sample. | 145 |
| **OLD SAMPLE:**<br>Sample greater than 15 days old when received at State Public Health Laboratory. | 25 |
| **DAMAGED SPECIMEN:**<br>Specimen damaged in transit. | 10 |
| **NO BLOOD:**<br>Filter paper submitted without blood. | 2 |
| **FILTER PAPER AND FORM BARCODES DO NOT MATCH:**<br>Bar code on filter paper does not match bar code on Newborn Screening Form. Collection forms contain barcodes on demographic, hearing and filter paper portions. The barcodes may not be altered in any way. If incorrect baby is sampled, do not remove filter paper and attach to a different demographic portion. If a sampling error occurs, the entire form needs to be voided and sample needs to be recollected on a new form. All barcodes must match laboratory copy, submitter copy, newborn hearing screen, and filter paper. | 7 |

| | |
|---|---|
| **MISSING, INCOMPLETE OR CONFLICTING PATIENT INFORMATION:**<br>Missing, incomplete or conflicting demographic information. | 14 |
| **SERUM RINGS:**<br>Serum separated into clear rings around blood spot. Possible causes: card dried vertically (on side) instead of flat; squeezing excessively around puncture site; allowing filter paper to come in contact with alcohol, hand lotion, etc. | 39 |
| **BLOOD ON OVERLAY COVER:**<br>Overlay cover came in contact with wet blood sample. Possible causes: sample is poor quality status because blood soaked from back of filter paper onto the gold colored backing of the form. The filter paper circles are designed to hold a specific quantity of blood. If the wet filter paper is allowed to come into contact with the paper backing of form, blood can be drawn out of filter making the quantitative tests performed by the Newborn Screening Laboratory invalid. It is very important that the wet filter paper does not come into contact with any surface until completely dry. | 8 |
| **OLD FORM:**<br>Sample received on out-of-date form. | 7 |
| **Total Poor Quality Samples Received** | 2,128<br>(2.31%) |

# Appendix 6: Newborn Blood Spot Screening Hemoglobinopathy Report 2016

**Specimens Received:**

| | | |
|---|---|---|
| Initial: | 74,542 | (80.8%) |
| Repeat: | 15,624 | (16.9%) |
| Poor Quality: | 2,128 | ( 2.3%) |
| **Total:** | **92,294** | |

| **Significant Results = 1,678** | | | | | |
|---|---|---|---|---|---|
| **Sickle Cell Disease** | | **Other Disease Conditions** | | **Trait Conditions** | |
| FS | 17 | FCA | 1 | FAS | 1,114 |
| FSA | 5 | FCX | 1 | FSAINC | 7 |
| FSC | 6 | FE | 2 | FAC | 298 |
| FC | 2 | Highly Elevated Barts | 1 | FCAINC | 2 |
| | | | | FAE | 44 |
| | | | | FAD | 39 |
| | | | | FAX | 136 |
| | | | | FASX | |
| | | | | FACX | |
| | | | | Slightly Elevated Barts | 3 |
| | | | | Other Trait condition | |
| **Total** | **30** | **Total** | **5** | **Total** | **1,643** |

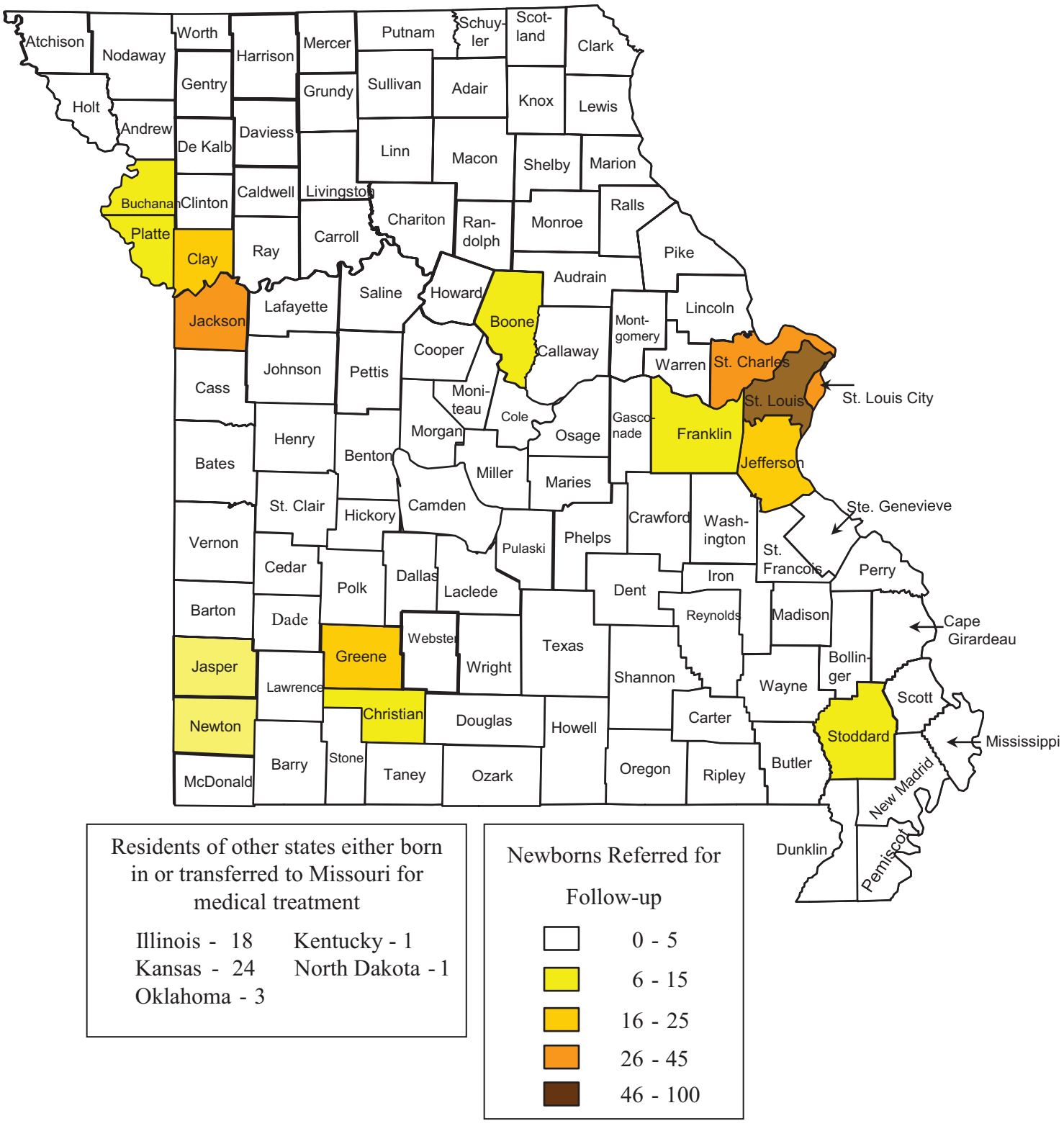
# Appendix 7: Newborn Hearing Screening Data for 2016

2016 calendar year provisional data for Missouri shows:

- 75,863 occurrent births (source: Department of Health and Senior Services Vital Records)
- 75,747 occurrent births (source: Missouri Health Strategic Architectures and Information Cooperative [MOHSAIC]*)
- 98.1% (74,349) of newborns were screened
- 98% (72,869) of infants were screened by 1 month of age
- 1.51% (1,151) of infants failed the final screening
- 90.2% (633) of the infants who failed their final screening and received an audiologic evaluation were evaluated and diagnosed by 3 months of age
- 100 infants were diagnosed with a permanent hearing loss

*The difference of 116 births between the occurrent birth count in the program data management system, the Missouri Health Strategic Architectures Information Collaborative (MOHSAIC), and the total occurrent births reported by Vital Records is the result of records that do not yet have an assigned Department Client Number (DCN) and records that are sealed. Records are not released from the Vital Records system to MOHSAIC until the DCN assignment is complete. Non-complete records are due to issues such as paternity and adoptions. Sealed birth records are neither displayed nor counted in MOHSAIC. This report is based upon MOHSAIC records.

# Appendix 8: Number of Newborns with Abnormal Newborn Blood Spot Screens Referred for Follow-up by County in 2016



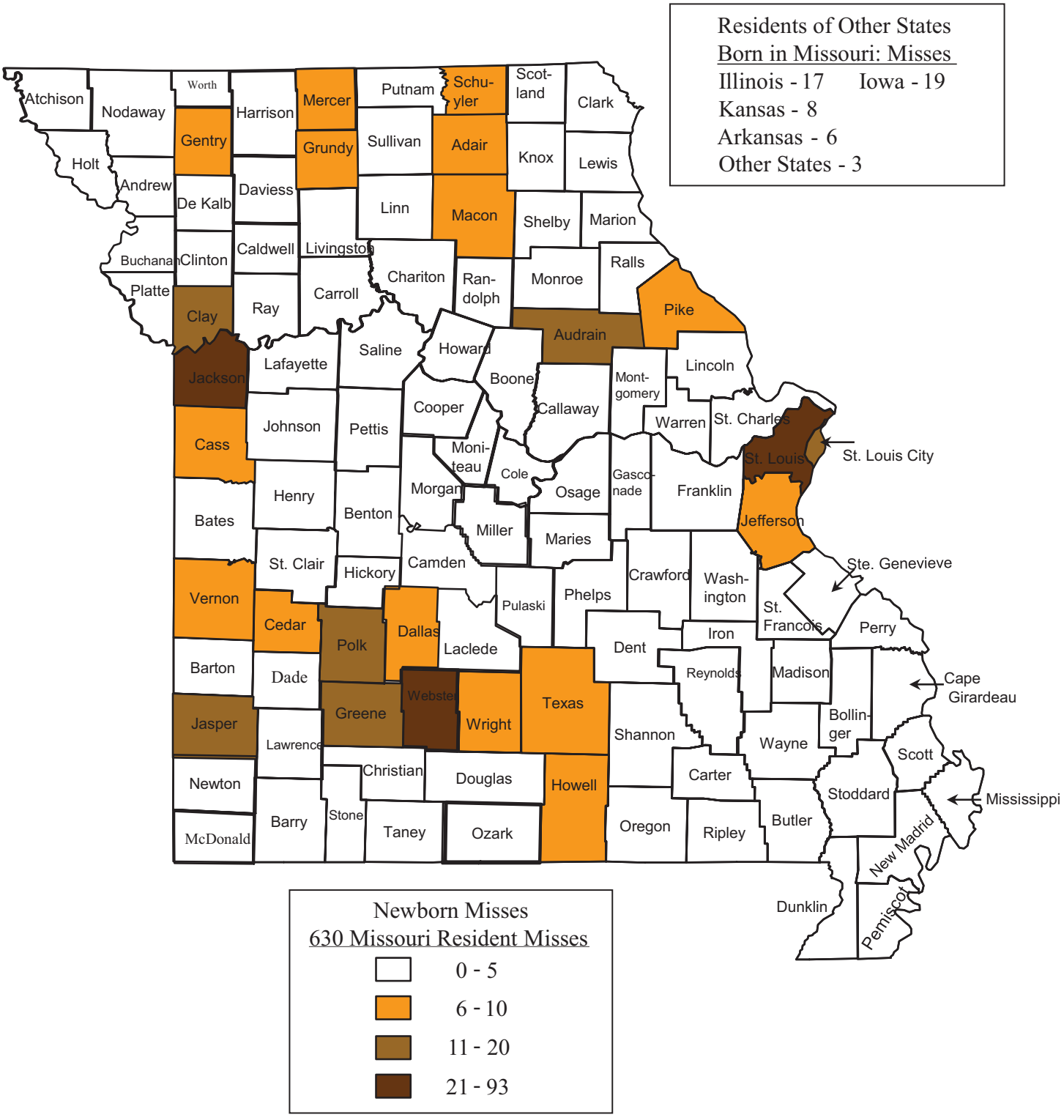

Residents of other states either born in or transferred to Missouri for medical treatment

| | |
|---|---|
| Illinois - 18 | Kentucky - 1 |
| Kansas - 24 | North Dakota - 1 |
| Oklahoma - 3 | |

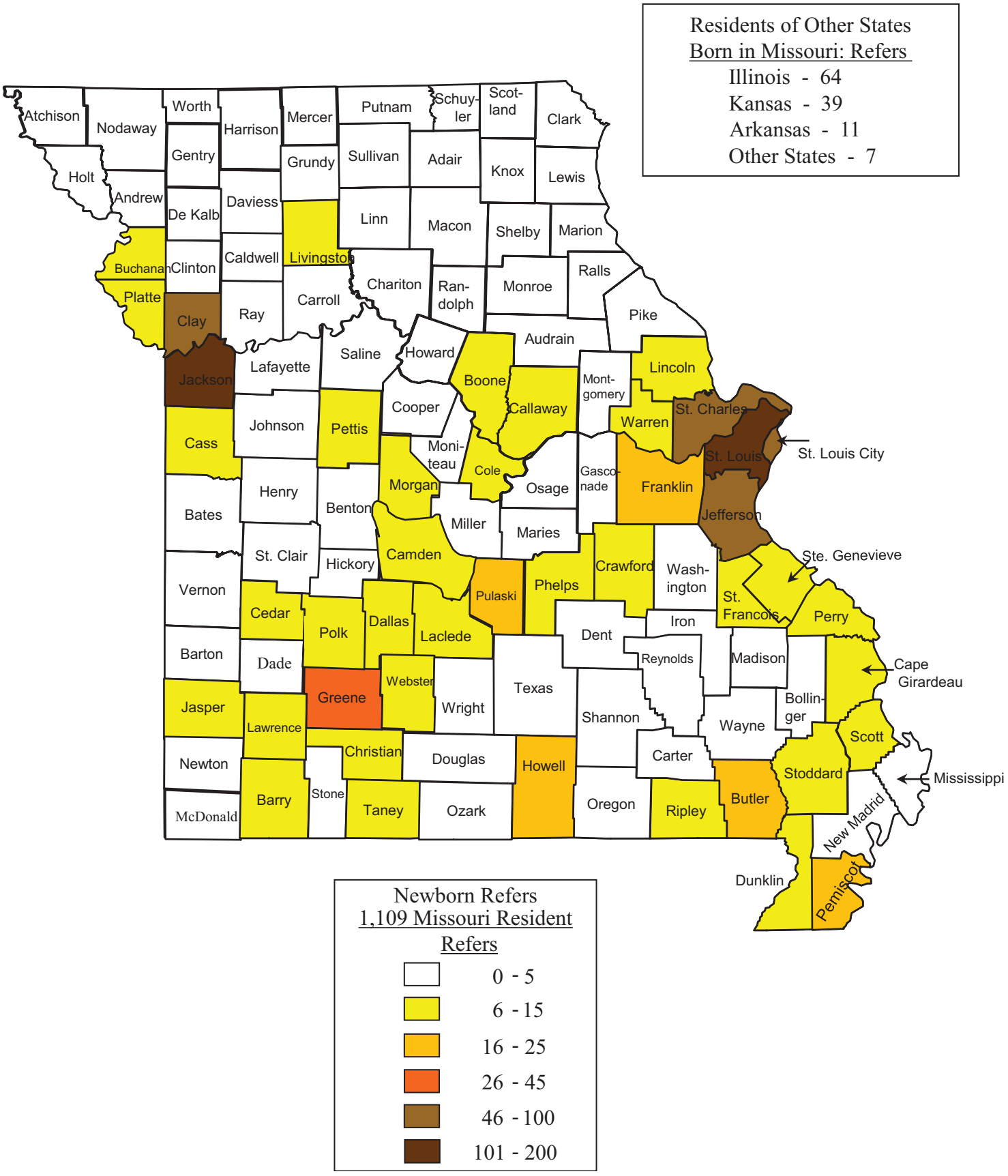
Newborns Referred for Follow-up

| | |
|---|---|
| | 0 - 5 |
| | 6 - 15 |
| | 16 - 25 |
| | 26 - 45 |
| | 46 - 100 |

# Appendix 9: Number of Newborns that Missed a Hearing Screening by County in 2016



# Appendix 10: Number of Newborns Referred After a Hearing Screen by County in 2016



# Appendix 11: Newborn Screening Parent Satisfaction Survey

A satisfaction survey of parents was conducted for families of babies having abnormal newborn screening results reported in 2016 and were confirmed positive. There were 154 satisfaction surveys* mailed and 19 were returned for a survey return rate of 12%. Key findings:

| Newborn Screening Parent Satisfaction Survey | | | |
|---|---|---|---|
| | Very Satisfied | Satisfied | Not Satisfied |
| Staff explained my baby's condition in a way I could understand. | 63% | 21% | 16% |
| Able to ask questions and discuss decisions about my baby's health care. | 74% | 16% | 10% |
| Offered reassurance and support. | 79% | 16% | 5% |
| The treatment staff was knowledgeable. | 84% | 16% | |
| My questions and concerns were addressed in a timely manner. | 68% | 16% | 16% |
| The staff provided me with useful referrals and resources. | 74% | 16% | 10% |
| Received high quality care during my appointments. | 74% | 21% | 5% |

Reasons parents responded as not satisfied include the length of time to see a specialist, primary care providers not being knowledgeable about the abnormal result the baby had, conflicting messages among center staff, and length of time it took to have laboratory results reported to the family.

*The number of surveys (154) sent to parents does not equal the number of confirmed positives (185) due to the following reasons: some parents had moved and surveys were returned with no forwarding address, some surveys were returned after the closing date of when the survey was to be returned, and surveys were not sent to parents whose child was not diagnosed within six months of the abnormal newborn screen. Also, this survey was not sent to parents whose child has sickle cell disease. Parents whose child has sickle cell disease were sent a separate survey whose results are stated on page 23.

A satisfaction survey of parents and children receiving services provided by the hemoglobinopathy resource centers was completed in 2016. There were 1,065 surveys mailed and 340 were returned for a survey return rate of 32%. Key findings:

| **Hemoglobinopathy Resource Center Satisfaction Survey - Parent Response** | | | |
|---|---|---|---|
| | Very Satisfied | Satisfied | Not Satisfied |
| Treated with respect | 97% | 1% | 2% |
| Treatment staff was knowledgeable | 88% | 12% | 0% |
| Questions/concerns addressed in a timely manner | 86% | 13% | 1% |
| Staff provided useful referrals and resources | 83% | 15% | 2% |
| Provided with the services needed | 97% | 2% | 1% |
| Medical care/services received | 76% | 23% | 1% |
| Received services or treatment without experiencing any problems | 97% | 0% | 3% |

Reasons parents responded as not satisfied with services were because of a long wait time. Parents did not indicate what a long wait time meant to them.

## Appendix 12: Newborn Hearing Screening Parent Satisfaction Survey

In February 2016*, a 2015 satisfaction survey was mailed to parents of children born in Missouri who failed their initial newborn hearing screening between October 2015 and December 2015. There were 517 surveys mailed and 84 were returned for a survey return rate of 16%. The survey examined factors influencing the follow-up time between a failed newborn hearing screening and a repeat screening or an audiologic evaluation.

Key findings:

- 74% of the respondents reported that the birth hospital provided them with written information about the hearing screening prior to the hearing screening.
- 99% of the respondents reported that the birth hospital notified them of the screening result.
- 80% of the respondents reported that the hospital staff explained the importance of knowing whether a baby has a hearing loss early in life.

*Survey conducted every two years.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